Molecular characterization of a Han Chinese family with essential hypertension

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Received November 18, 2015
Accepted January 7, 2016
Published May 13, 2016
DOI http://dx.doi.org/10.4238/gmr.15028084

ABSTRACT. Mutations in the mitochondrial genome have been found to be associated with essential hypertension. Here, we report the clinical and molecular characterization of a three-generation Han Chinese family with maternally inherited hypertension. Most strikingly, this pedigree exhibited a high penetrance of hypertension. Sequence analysis of the mitochondrial genome showed the presence of a homoplasmic T16189C mutation in the D-loop and the intergenic CO2/tRNA<sup>Lys</sup> 9-bp common deletion, as well as a set of polymorphisms belonging to the East Asia haplogroup B5b1. The well-known T16189C mutation, which is in the first hypervariable segment of the mitochondrial control region, is implicated to be associated with a wide range of clinical disorders. Moreover, the genetic polymorphism 9-bp common deletion is found to be associated with hepatocellular carcinoma in the Han Chinese population. Thus, the combination of T16189C mutation and the 9-bp deletion may have caused mitochondrial dysfunction and contributed to the development of essential hypertension in this Chinese family.

Key words: Hypertension; mtDNA; Mutation; Chinese family